Types of EB

Epidermolysis Bullosa (EB)

EB-1
- Inherited EB
- EB hereditaria

- Mechano-bullous → tendency to develop blisters following minor trauma / friction of skin

Causes

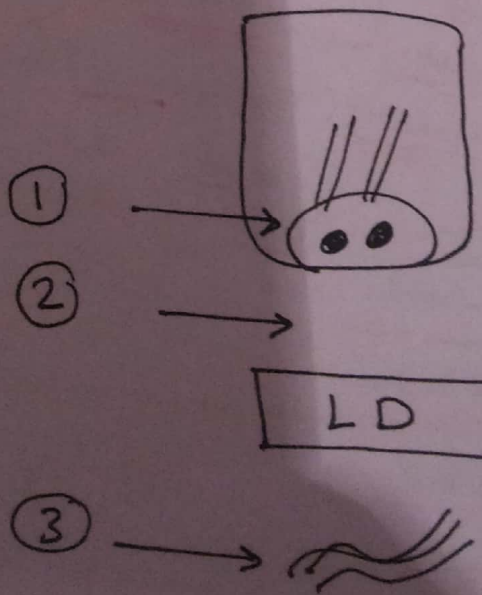
- Mutations in the genes encoding proteins of DEJ → sub-epidermal or intraepidermal (15 structural protein)
- According to
  - mutated gene
  - mode of inheritance
  - defective protein
  - level of blister

} 3 main types

Types of EB (1)

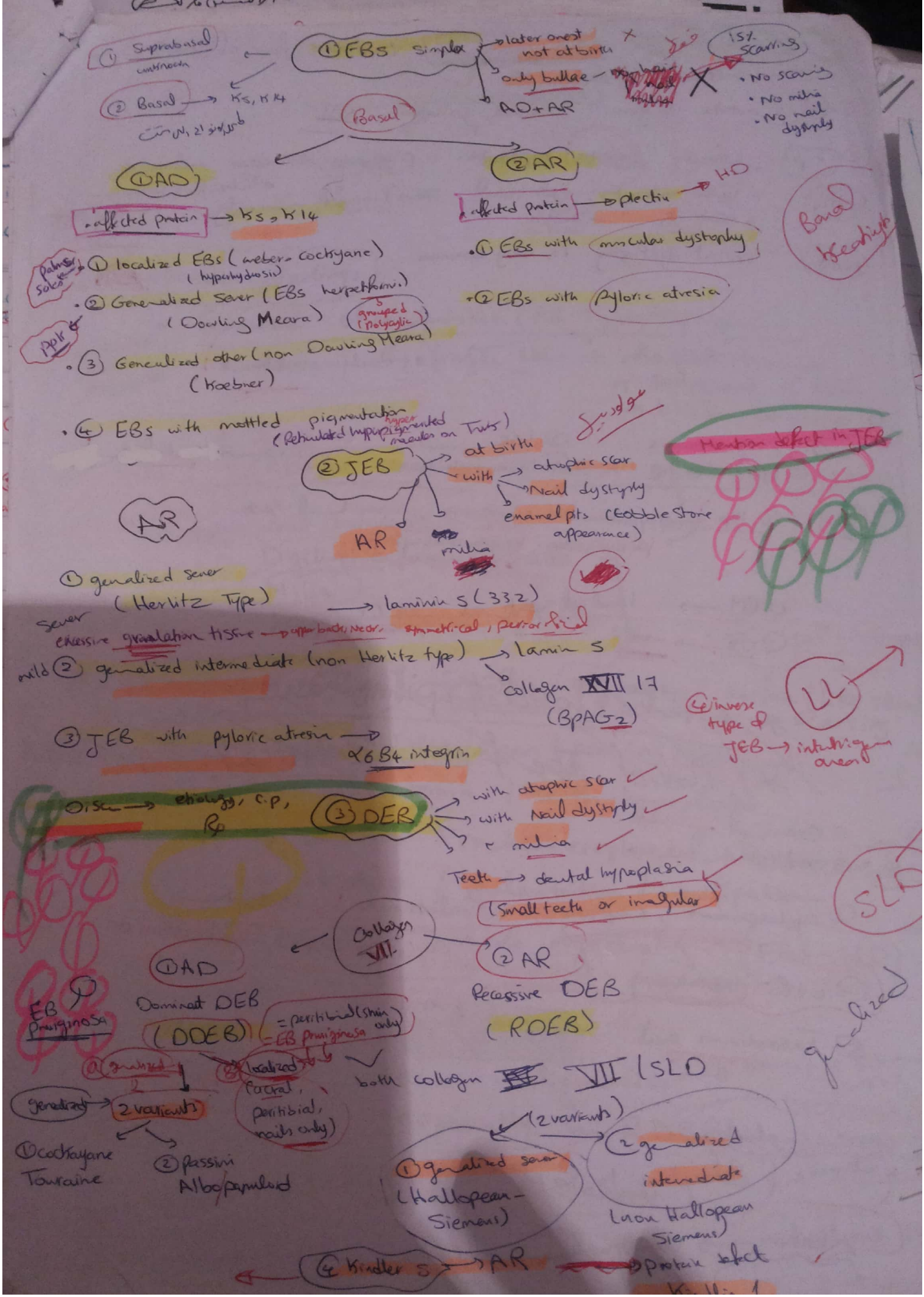
EB
- Immunobullous → EBA
- Genetic → EB hereditaria

EB (4)
1. Simplex (intraepidermal)
   - Basal KCs in BMZ
   - (1)
   - Target Ag: K5, 14
2. Junctional
   - LL
   - (2)
3. Dystrophic
   - SLD
   - (3)



Update:
4 types
1. simplex
2. JEB
3. DEB
4. Kindler's $

(4)



EB-3

Clinical picture

Cutaneous → Extra-cutaneous

EB
- at birth
- Trauma induced (at sites of Trauma)
- +ve Family history

① * Cutaneous findings

- General characteristics (All types/subtypes):

Epidermolysis bullosa Acquisita

① Mechanically fragile skin → minimal trauma or movement of skin → blister formation

② Erosions

③ Atrophic scarring (more in DEB & JEB > SEB)
DEB 100%, SEB 15%

EBS → no scarring, No Nail, No hair affected

Question: nail changes in EBD
- Dystrophic / absent nails (× EBS)
- Milia (× EBS, EBJ)
- Scarring alopecia of scalp

→ more RDEB, lesser extent → JEB, EBS with pyloric atresia, muscular dystrophy

② * Extracutaneous findings — الأغشية: any mucosal surface (like skin) → erosions → scarring

Defect in protein (Basement membrane) all over the body

- Eye → Corneal blisters / ulcers / scarring / Ectropion
- Oral cavity → microstomia / teeth hypoplasia
- Upper resp. tract → Tracheo-esophageal stenosis → dysphagia, dyspnea
- GIT → Strictures / malnutrition / Constipation (EBS with PA, JEB with PA)
- Musculo-skeletal → muscle dystrophy (EBS-MD)
  - Pseudosyndactyly → digits totally fused by scar tissue (mitten) deformity of hands/feet
  - osteoporosis
  - osteopenia

Extracutaneous complications more with EBD → more scarring

EBD > JEB > SEB

③ Complications

Skin malignancies → on top of chronic scarring, non healing wounds

especially in RDEB → especially in severe generalized type (Hallopeau Siemens)

(SCC - Melanoma)

① well differentiated
② frequently metastasize
③ c/p → indistinct borders (difficult to completely excised surgically)
④ no response to radio, chemo

↓ leading cause of death (4-5 days)

History → age
→ family history
Trauma

Not useful → may be useful in very superficial form of EB

EB-4

Diagnosis
- → EM → standard diagnostic Method — Transmission EM
- → Genetic study & IF Antigenic mapping → To detect the affected gene or protein

① L/M (routine histopathology) : NOT useful
- × Can't differentiate level of cleft < LL / SLD
- × may be useful in very superficial types of EBS

② E/M (standard diagnostic procedure)
- → Transmission EM (TEM)

③ Genetic studies / IF (Immunofluorescence) antigenic mapping ? — ①② gold standard
- to determine the defective gene / protein

4) molecular diagnosis

. No Role to Histopathology.

D.D. EB [illegible]

Mechanobullous disease →
- chronic
- early onset
- +ve family history
- -ve DIF

Trauma / infection

- vesicles & bullae in neonates → HSV, SSSS, Bullous impetigo, Bullous mastocytosis
- nail & teeth affection: → pachyonychia congenita
  →
- History

DD ① Bullae in neonates
- ① Trauma → Suckling Blister
- ② infection → HSV, SSSS, Bullous impetigo
- ③ Genetic → ① Bullous congenital ichthyosiform erythroderma, ② incontinentia pigmenti (vesicular stage) ③ EB < S, D, Junctional
- ④ Tumors → Bullous mastocytosis → Blisters → erosion, crustation
- ⑤ Metabolic → acrodermatitis
- ⑥ autoimmune → Pemphigoid gestationis, neonatal pemphigus acquired transplacentally
- ⑦ milliaria cystallina

Treatment of EB

EB-5

There's No specific TR for any type of EB

General measures

- Guard against:
  - trauma
    - loose clothes
    - pads over bony prominence
  - infection
    - anti-septic lotions
    - antibiotics

Skin care (dressings)

non-adhesive

Systemic drugs

Topical phenytoin.

- phenytoin ⊖ Collagenases ↑ Collagen
- Human collagen injection

Healsol / Healsol Spray

- Fibroblast activity → ↑ fibrosis

others

- Gene therapy → IO injection of collagen VII (7)

Chemoprophylaxis

Isotretanoin → Suppress Collagenase & may cause Hypertrophic scar (Prevent SCC)

Low dosis but may ↑ mechanical fragility / Blistering of SK.

Colchicin → ⊕ Collagenase so used in Rx of Syst. sclerosis

cyclosporine and Thalidomide for symptomatic relief of EB Pruriginosa

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P185

localized form of EBS

EB pruriginosa

- rare subtype of dystrophic EB
- ① defect: mutation in gene coding collagen VII
- ② C.P: intensely pruritic hypertrophic plaques in linear configuration
  - lower extremities (pretibial skin) - most commonly
  - thickened urticaria papule, nodule plaques
- Skin
  - scarring + milia
- Nail
  - Nail dystrophy is very common

shortly after birth ↙ ↘ or → in early adulthood

③ OO: ① prurigo nodularis ② hypertrophic L.P ③ LSC ④ lichen amyloidosis

⑤ Dermatitis artefacta

④ Dicg: ① Hp → hyperkeratosis, acanthosis, subepidermal fibrosis
perivascular lymphohistiocytic

② EM → level of split at SLD

③ DF → -ve

* Orf florid Papillomatosis
* ~~Florid Type~~ of endemic African ~~Kaposi's~~ Sa

⑤ ① General →
⑥ ② antihistamine →

② cyclosporine, Dapsone, Thalidomide, isotretenoin
cyclosporine + thalidomide

③ UVB phototherapy → photo

⑤ Tacrolimus oint.

chronic

⑥ Course, complication
① Chronic, Refractory to Rx
② Complication → Lymphoedema → SCC ??? dll

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